

Isovaleric Acidemia (IVA)

An organic acid disorder

What is it?

Isovaleric Acidemia (also known as IVA) is an inherited organic acid disorder. People with organic acid disorders, like IVA, cannot properly break down certain components of protein. This is because the body is lacking a specific chemical called an enzyme. Since the body cannot properly break down the protein, certain organic acids build up in the blood and urine and cause problems when a person eats normal amounts of protein, or becomes sick.

What are the symptoms?

A person with IVA can appear normal at birth. Some people with IVA will have the following symptoms after a few days of life: poor feeding, lack of energy, vomiting, and have a smell like sweaty feet. There is an increased risk for infection. Some people with IVA may not have symptoms until later in infancy or childhood. Symptoms are usually associated with an illness, like a virus, cold, or the flu, or when high-protein foods are eaten. Many symptoms of IVA can be prevented by immediate treatment and lifelong management. People with IVA typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

Inheritance and frequency

IVA is inherited in an autosomal recessive manner. This means that for a person to be affected with IVA, he or she must have inherited two non-working copies of the gene responsible for causing IVA. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non working copies of their gene, causing the baby to have IVA. Typically, there is no family history of IVA in an affected person. IVA is a rare fatty acid oxidation disorder; about 1 in 200,000 babies born have IVA.

How is it detected?

IVA may be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

How is it treated?

IVA is treated by eating a low protein diet and drinking a special formula, and sometimes medication, as recommended by a genetic metabolic medical professional.

DISCLAIMER: This information is not intended to replace the advice of a genetic metabolic medical professional.

For more information:

Genetics Home Reference

Website: <http://www.ghr.nlm.nih.gov>

Save Babies Through Screening Foundation

4 Manor View Circle

Malvern, PA 19355-1622

Toll Free Phone: 1-888-454-3383

Fax: (610) 993-0545

Email: email@savebabies.org

Website: <http://www.savebabies.org>

Organic Acidemia Association

13210 - 35th Avenue North

Plymouth, MN 55441

Phone: 763-559-1797

Fax: 763-694-0017

Email: oaanews@aol.com

www.oaanews.org